

Carnitine Palmitoyltransferase I (CPT I) Deficiency

4-18 years

Clinic Visits:

- At least every year

Laboratory studies:

- Clinic visits (yearly)
 - Comprehensive metabolic panel
 - Urine analysis
 - Triglycerides
 - Phosphate (blood and urine)
 - Urine creatinine

During illness:

- Call the Metabolic Clinic to alert them immediately if your child is sick
 - Poor feeding, vomiting, and/or lethargy
- Give your child glucose gel or polymer solution if needed (or instructed to do so)
- If your child is unable to keep down glucose containing fluids, you will need to go to the emergency room for IV 10% dextrose (glucose)
- Be sure to bring Emergency Room letter with you to the hospital
- Labs during illness include: comprehensive metabolic panel, ammonia, serum phosphate, and serum CK (muscle health)
- Please alert the Metabolic Clinic if your child need surgery
- Wear Medic Alert Bracelet!!!**

Evaluations:

- Yearly developmental questionnaires to be completed by parents
- Developmental evaluation at age 6 years
- Neuropsychological evaluation at age 9 years

Topics:

- Review of biochemistry and genetics of CPT I deficiency
- Diet/management
 - Avoid fasting
 - Management during illness
 - Possible medium chain triglyceride (MCT) oil

Please make sure your child has clinic visits and laboratory studies done on time. Call the metabolic clinic at _____ to make an appointment, schedule a blood draw, or with any other questions. If you urgently need to reach a metabolic physician, call _____ and ask to page the metabolic physician on call.

Thank You,
Your Metabolic Team